Connecting via Winsock to Dialog

Logging in to Dialog

Trying 31060000009999...Open

DIALOG INFORMATION SERVICES
PLEASE LOGON:
****
ENTER PASSWORD:
****
Welcome to DIALOG

Dialog level 02.16.02D

Last logoff: 01jul03 18:05:19
Logon file405 02jul03 11:44:28
*** ANNOUNCEMENT ***
***
--File 654 - US published applications from March 15, 2001 to the present are now online. Please see HELP NEWS 654 for details.

***
--File 581 - The 2003 annual reload of Population Demographics is complete. Please see Help News581 for details.

***
--File 156 - The 2003 annual reload of ToxFile is complete. Please see HELP NEWS156 for details.

***
--File 990 - NewsRoom now contains February 2003 to current records.
File 992 - NewsRoom 2003 archive has been newly created and contains records from January 2003. The oldest months's records roll out of File 990 and into File 992 on the first weekend of each month.
To search all 2003 records BEGIN 990, 992, or B NEWS2003, a new OneSearch category.
***
--Connect Time joins DialUnits as pricing options on Dialog.
See HELP CONNECT for information.
***
***
--SourceOne patents are now delivered to your email inbox as PDF replacing TIFF delivery. See HELP SOURCE1 for more information.
***
--Important news for public and academic libraries. See HELP LIBRARY for more information.
***
--Important Notice to Freelance Authors--
See HELP FREELANCE for more information
***
NEW FILES RELEASED
***World News Connection (File 985)
***Dialog NewsRoom - 2003 Archive (File 992)
***TRADEMARKSCAN-Czech Republic (File 680)
***TRADEMARKSCAN-Hungary (File 681)
***TRADEMARKSCAN-Poland (File 682)
***
UPDATING RESUMED

***

RELOADED
***Population Demographics -(File 581)
***CLAIMS Citation (Files 220-222)

REMOVED
***

>>> Enter BEGIN HOMEBASE for Dialog Announcements <<<
>>> of new databases, price changes, etc. <<<
****

* * * * See HELP NEWS 225 for information on new search prefixes and display codes

*** ***

SYSTEM:HOME
Cost is in DialUnits
Menu System II: D2 version 1.7.9 term=ASCII
*** DIALOG HOMEBASE(SM) Main Menu ***

Information:
1. Announcements (new files, reloads, etc.)
2. Database, Rates, & Command Descriptions
3. Help in Choosing Databases for Your Topic
4. Customer Services (telephone assistance, training, seminars, etc.)
5. Product Descriptions

Connections:
6. DIALOG(R) Document Delivery
7. Data Star(R)

/H = Help /L = Logoff /NOMENU = Command Mode

Enter an option number to view information or to connect to an online service. Enter a BEGIN command plus a file number to search a database (e.g., B1 for ERIC).
? b 410

02jul03 11:44:29 User268147 Session D98.1
$0.00 0.149 DialUnits FileHomeBase
$0.00 Estimated cost FileHomeBase
$0.00 Estimated cost this search
$0.00 Estimated total session cost 0.149 DialUnits

File 410:Chronolog(R) 1981-2003/Aug
(c) 2003 The Dialog Corporation

Set Items Description
--- ---- ---------
? set hi %%%;set hi %%%
HILIGHT set on as "
HILIGHT set on as "
? b 5, 34, 155, 172
02jul03 11:44:40 User268147 Session D98.2
$0.00 0.071 DialUnits File410
$0.00 Estimated cost File410
$0.04 TELNET
$0.04 Estimated cost this search

```
   $0.04  Estimated total session cost  0.220 DialUnits

SYSTEM:OS  - DIALOG OneSearch
 File  5:Biosis Previews(R) 1969-2003/Jun W4
      (c) 2003 BIOSIS
 File 34:SciSearch(R) Cited Ref Sci 1990-2003/Jun W5
      (c) 2003 Inst for Sci Info
 File 155:MEDLINE(R) 1966-2003/Jun W4
      (c) format only 2003 The Dialog Corp.
*File 155: Medline has been reloaded and accession numbers have
changed.  Please see HELP NEWS 155.
 File 172:EMBASE Alert 2003/Jun W5
      (c) 2003 Elsevier Science B.V.

    Set  Items  Description
    ---  -----  ---------
? s "epidermolysis bullosa"
    S1   1899  "EPIDERMOLYSIS BULLOSA"
? s cytosine
    S2  48128  CYTOSINE
? s 1368
    S3    586  1368
? s ?1368?
>>>File 5 processing for ?1368? stopped at ACROLAXUS ORIENTALIS (ORGANISMS
  - UNSPECIFIED)
>>>File 34 processing for ?1368? stopped at ADDNRHOK□
>>>File 155 processing for ?1368? stopped at ALLERGOGENI
□
    S4      0  ?1368?
? s 1368?
    S5    700  1368?
? s laminin
    S6  40863  LAMININ
? s s6 and (insert or mutation or mutated or insertion or deletion or substitution)
       40863  S6
       29972  INSERT
      522151  MUTATION
       60560  MUTATED
      167009  INSERTION
      221200  DELETION
      206582  SUBSTITUTION
    S7   1810  S6 AND (INSERT OR MUTATION OR MUTATED OR INSERTION OR
           DELETION OR SUBSTITUTION)
? ds

Set    Items  Description
S1      1899  "EPIDERMOLYSIS BULLOSA"
S2     48128  CYTOSINE
S3       586  1368
S4         0  ?1368?
S5       700  1368?
S6     40863  LAMININ
S7      1810  S6 AND (INSERT OR MUTATION OR MUTATED OR INSERTION OR DELE-
         TION OR SUBSTITUTION)
? s s3 or s5
       586  S3
       700  S5
    S8   700  S3 OR S5
? s s1 and s8
      1899  S1
       700  S8
    S9     0  S1 AND S8
```

1-2
2-6
5-6
7.

? s s8 and s2
700 S8
48128 S2
S10 0 S8 AND S2
?
PLEASE ENTER A COMMAND OR BE LOGGED OFF IN 5 MINUTES
? s s7 ands1
>>>Term "ANDS1" in invalid position
? s s7 and s1
1810 S7
1899 S1
S11 37 S7 AND S1
? s s11 and cytosine
37 S11
48128 CYTOSINE
S12 1 S11 AND CYTOSINE
? type s12/full/all

12/9/1 (Item 1 from file: 34)
DIALOG(R)File 34:SciSearch(R) Cited Ref Sci

11469061 Genuine Article#: 654YK Number of References: 22
Title: A mutation in the LAMC2 gene causes the Herlitz junctional epidermolysis bullosa (H-JEB) in two French draft horse breeds
Author(s): Milenkovic D; Chaffaux S; Taourit S; Guerin G (REPRINT)
Corporate Source: INRA,Ctr Rech Jouy, Dept Genet Anim, Lab Genet Biochim & Cytogenet,F-78352 Jouy En Josas//France/ (REPRINT); INRA,Ctr Rech Jouy, Dept Genet Anim, Lab Genet Biochim & Cytogenet,F-78352 Jouy En Josas//France/
Journal: GENETICS SELECTION EVOLUTION, 2003, V35, N2 (MAR-APR), P249-256
ISSN: 0999-193X Publication date: 20030300
Publisher: E D P SCIENCES, 7, AVE DU HOGGAR, PARC D ACTIVITES COURTABOEUF, BP 112, F-91944 LES ULIS CEDEXA, FRANCE
Language: English Document Type: ARTICLE
Geographic Location: France
Journal Subject Category: AGRICULTURE, DAIRY & ANIMAL SCIENCE; GENETICS & HEREDITY
Abstract: Epidermolysis bullosa (EB) is a heterogeneous group of inherited diseases characterised by skin blistering and fragility. In humans, one of the most severe forms of EB known as Herlitz-junctional EB (H-JEB), is caused by mutations in the laminin 5 genes. EB has been described in several species, like cattle, sheep, dogs, cats and horses where the mutation, a cytosine insertion in exon 10 of the LAMC2 gene, was very recently identified in Belgian horses as the mutation responsible for JEB. In this study, the same mutation was found to be totally associated with the JEB phenotype in two French draft horse breeds, Trait Breton and Trait Comtois. This result provides breeders a molecular test to better manage their breeding strategies by genetic counselling.
Descriptors--Author Keywords: horse ; LAMC2 ; epidermolysis bullosa ; laminin 5
Identifiers--KeyWord Plus(R): MECHANOBULLOUS DISEASE; CLASSIFICATION; DIAGNOSIS; POSITION
Cited References:
AUMAILLEY M, 1998, V193, P1, J ANAT 1
BRENNEMAN KA, 2000, V37, P4336, VET PATHOL
BRETHELSEN H, 1935, V48, P258, J COMP PATH THER
BRUCKNERTUDERMA.L, 1991, V96, P452, J INVEST DERMATOL
COLOGNATO H, 1999, V9, P1327, CURR BIOL
CROWELL WA, 1976, V168, P56, J AM VET MED ASSOC
DUBIELZIG RR, 1986, V23, P325, VET PATHOL

FINE JD, 2000, V42, P1051, J AM ACAD DERMATOL
FINE JD, 1991, V24, P119, J AM ACAD DERMATOL
FRAME SR, 1988, V193, P1420, J AM VET MED ASSOC
GOUREAU JM, 1989, V62, P345, B ACAD VET FR
HOOD J, 2001, V11, P463, TRENDS CELL BIOL
JOHNSON GC, 1998, V99, P329, J COMP PATHOL
KOHN CW, 1989, V21, P297, EQUINE VET J
KORGE BP, 1996, V74, P59, J MOL MED-JMM
LYKKEANDERSEN J, 2001, V293, P1836, SCIENCE
NAGY E, 1998, V23, P198, TRENDS BIOCHEM SCI
OLIVRY T, 1999, V36, P616, VET PATHOL
PALAZZI X, 2000, V115, P135, J INVEST DERMATOL
PULKKINEN L, 1999, V18, P29, MATRIX BIOL
SPIRITO F, 2002, V3, P684, J INVEST DERMATOL
TERWILLIGER JD, 1995, V56, P777, AM J HUM GENET

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? s lamc2
   S13    201  LAMC2
? ds

Set    Items  Description
S1      1899  "EPIDERMOLYSIS BULLOSA"
S2     48128  CYTOSINE
S3       586  1368
S4         0  ?1368?
S5       700  1368?
S6     40863  LAMININ
S7      1810  S6 AND (INSERT OR MUTATION OR MUTATED OR INSERTION OR DELE-
         TION OR SUBSTITUTION)
S8       700  S3 OR S5
S9         0  S1 AND S8
S10        0  S8 AND S2
S11       37  S7 AND S1
S12        1  S11 AND CYTOSINE
S13      201  LAMC2
? s s13 and s3
         201  S13
         586  S3
   S14     0  S13 AND S3
? s s13 and s5
         201  S13
         700  S5
   S15     3  S13 AND S5
? type s15/full/all

 15/9/1    (Item 1 from file: 5)
DIALOG(R)File  5:Biosis Previews(R)
(c) 2003 BIOSIS. All rts. reserv.

13909810  BIOSIS NO.: 200200538631
Animal models for skin blistering conditions: Absence of laminin 5 causes
 hereditary junctional mechanobullous disease in the Belgian horse.
AUTHOR: Spirito Flavia; Charlesworth Alexandra; Linder Keith; Ortonne
 Jean-Paul; Baird John; Meneguzzi Guerrino(a)
AUTHOR ADDRESS: (a)INSERM U385, UFR de Medecine, Avenue de Valombrose,
 06107, Nice Cedex 2**France E-Mail: meneguzz@unice.fr
JOURNAL: Journal of Investigative Dermatology  119 (3):p684-691 September,
2002
MEDIUM: print
ISSN: 0022-202X
DOCUMENT TYPE: Article
RECORD TYPE: Abstract
LANGUAGE: English
```

ABSTRACT: Recent achievements in the genetic correction of keratinocytes isolated from patients with junctional epidermolysis bullosa have paved the way to a gene therapy approach for the disease. Because gene therapy protocols require preclinical validation in animals, we have characterized spontaneous animal models of junctional epidermolysis bullosa. In this study we have elucidated the genetic basis of the hereditary junctional mechanobullous disease in the Belgian horse, a condition characterized by blistering of the skin and mouth epithelia, and exungulation (loss of the hoof). Immunofluorescence analysis associated the condition to the absent expression of the gamma2 chain of laminin 5 and designated Lamc2 as the candidate gene. Comparative analysis of the nucleotide sequence of the full-length gamma2 cDNA isolated by reverse transcription polymerase chain reaction amplification of total RNA purified from the epithelium of a junctional epidermolysis bullosa foal and a healthy control disclosed a homozygous basepair insertion (1368insC) in the affected animal. Mutation 1368insC results in a downstream premature termination codon and is predicted to cause absent expression of the laminin gamma2 polypeptide. Our results also show that: (i) the horse junctional epidermolysis bullosa genetically corresponds to the severe Herlitz form of junctional epidermolysis bullosa in man; (ii) the amino acid sequence and structure of the horse laminin gamma2 chain are virtually identical to the human counterpart; (iii) the moderate eruption of skin blisters in the affected animals with respect to the human Herlitz junctional epidermolysis bullosa patients correlates with the protection provided by hair. Our observations suggest that the affected foals are a convenient source of epithelial cells from tissues that cannot be obtained from human junctional epidermolysis bullosa patients, and imply that hairless strains of animals with recessive skin disorders would be the best models for in vivo gene therapy approaches to skin blistering diseases.

DESCRIPTORS:
MAJOR CONCEPTS: Biochemistry and Molecular Biophysics; Integumentary System (Chemical Coordination and Homeostasis)
BIOSYSTEMATIC NAMES: Equidae--Perissodactyla, Mammalia, Vertebrata, Chordata, Animalia
ORGANISMS: horse (Equidae)--animal model, breed-Belgian, foal
ORGANISMS: PARTS ETC: epidermis--integumentary system; hoof--integumentary system; mouth epithelia--dental and oral system
BIOSYSTEMATIC CLASSIFICATION (SUPER TAXA): Animals; Chordates; Mammals; Nonhuman Mammals; Nonhuman Vertebrates; Perissodactyls; Vertebrates
DISEASES: epitheliogenesis imperfecta--integumentary system disease; exungulation--integumentary system disease; genodermatosis--integumentary system disease; hereditary junctional mechanobullous disease--genetic disease, integumentary system disease; skin blistering--integumentary system disease
CHEMICALS & BIOCHEMICALS: cDNA {complementary DNA}; laminin 5--absence
CONCEPT CODES:
03506 Genetics and Cytogenetics-Animal
10060 Biochemical Studies-General
10062 Biochemical Studies-Nucleic Acids, Purines and Pyrimidines
18504 Integumentary System-Physiology and Biochemistry
18506 Integumentary System-Pathology
19004 Dental and Oral Biology-Physiology and Biochemistry
BIOSYSTEMATIC CODES:
86145 Equidae

15/9/2 (Item 1 from file: 34)
DIALOG(R)File 34:SciSearch(R) Cited Ref Sci

11111016 Genuine Article#: 592RV Number of References: 52
Title: Animal models for skin blistering conditions: Absence of laminin 5 causes hereditary junctional mechanobullous disease in the Belgian horse
Author(s): Spirito F (REPRINT) ; Charlesworth A; Linder K; Ortonne JP; Baird J; Meneguzzi G
Corporate Source: Fac Med,INSERM U385, UFR Med,Ave Valombrose/F-06107 Nice 2//France/ (REPRINT); Fac Med,INSERM U385, UFR Med,F-06107 Nice 2//France/; Univ Guelph,Ontario Vet Coll, Dept Pathobiol,Guelph/ON N1G 2W1/Canada/; Univ Guelph,Ontario Vet Coll, Dept Clin Studies,Guelph/ON N1G 2W1/Canada/
Journal: JOURNAL OF INVESTIGATIVE DERMATOLOGY, 2002, V119, N3 (SEP), P 684-691
ISSN: 0022-202X Publication date: 20020900
Publisher: BLACKWELL PUBLISHING INC, 350 MAIN ST, MALDEN, MA 02148 USA
Language: English Document Type: ARTICLE
Geographic Location: France; Canada
Journal Subject Category: DERMATOLOGY & VENEREAL DISEASES
Abstract: Recent achievements in the genetic correction of keratinocytes isolated from patients with junctional epidermolysis bullosa have paved the way to a gene therapy approach for the disease. Because gene therapy protocols require preclinical validation in animals, we have characterized spontaneous animal models of junctional epidermolysis bullosa. In this study we have elucidated the genetic basis of the hereditary junctional mechanobullous disease in the Belgian horse, a condition characterized by blistering of the skin and mouth epithelia, and exungulation (loss of the hoof): Immunofluorescence analysis associated the condition to the absent expression of the gamma2 chain of laminin 5 and designated Lamc2 as the candidate gene. Comparative analysis of the nucleotide sequence of the full-length gamma2 cDNA isolated by reverse transcription polymerase chain reaction amplification of total RNA purified from the epithelium of a junctional epidermolysis bullosa foal and a healthy control disclosed a homozygous basepair insertion (1368insC) in the affected animal. Mutation 1368insC results in a downstream premature termination codon and is predicted to cause absent expression of the laminin gamma2 polypeptide. Our results also show that: (i) the horse junctional epidermolysis bullosa genetically corresponds to the severe Herlitz form of junctional epidermolysis bullosa in man; (ii) the amino acid sequence and structure of the horse laminin gamma2 chain are virtually identical to the human counterpart; (iii) the moderate eruption of skin blisters in the affected animals with respect to the human Herlitz junctional epidermolysis bullosa patients correlates with the protection provided by hair. Our observations suggest that the affected foals are a convenient source of epithelial cells from tissues that cannot be obtained from human junctional epidermolysis bullosa patients, and imply that hairless strains of animals with recessive skin disorders would be the best models for in vivo gene therapy approaches to skin blistering diseases.
Descriptors--Author Keywords: epitheliogenesis imperfecta ; genodermatosis ; Lamc2
Identifiers--KeyWord Plus(R): DYSTROPHIC EPIDERMOLYSIS-BULLOSA; CORRECTIVE GENE-TRANSFER; GAMMA-2 CHAIN; BRANCHING MORPHOGENESIS; MONOCLONAL-ANTIBODY; EPITHELIAL-CELLS; VII COLLAGEN; LAMB3 GENE; B2 CHAIN; EXPRESSION
Cited References:
ABERDAM D, 1994, V6, P299, NAT GENET
ABERDAM D, 1994, V2, P115, CELL ADHES COMMUN
AMANO S, 2000, V275, P22728, J BIOL CHEM
BERTHELSEN H, 1935, V48, P285, J COMP PATHOL THER
BRUCKNERTUDERMA.L, 1991, V96, P452, J INVEST DERMATOL

BUTZ H, 1957, V64, P555, DTSCH TIERARZTLICHE
CHAMPLIAUD MF, 1996, V132, P1189, J CELL BIOL
CHOATE KA, 1996, V7, P2247, HUM GENE THER
COOPER DN, 1993, V25, P7, ANN MED
CUI Y, 1995, V9, P423, GENE DEV
DELLAMBRA E, 1998, V9, P1359, HUM GENE THER
FINE JD, 2000, V42, P1051, J AM ACAD DERMATOL
FRAME SR, 1988, V193, P1420, J AM VET MED ASSOC
FREIBERG RA, 1997, V6, P927, HUM MOL GENET
GACHE Y, 1996, V97, P2289, J CLIN INVEST
GAGNOUXPALACIOS L, 2001, V153, P835, J CELL BIOL
GEDDEDAHL T, 1996, P1225, EMERY RIMOINS PRINCI
GHAZIZADEH S, 1999, V6, P1267, GENE THER
GOURREAU JM, 1990, V22, P65, POINT VET
HEINONEN S, 1999, V112, P3641, J CELL SCI
HORMIA M, 1998, V77, P1479, J DENT RES
JOHNSON GC, 1988, V98, P329, J COMP PATHOL
KADOYA Y, 1999, V112, P417, HISTOCHEM CELL BIOL
KALLUNKI P, 1992, V119, P679, J CELL BIOL
KOHN CW, 1989, V21, P297, EQUINE VET J
KUSTER JE, 1997, V8, P673, MAMM GENOME
MARINKOVICH MP, 1992, V267, P17900, J BIOL CHEM
MATSUI C, 1995, V105, P648, J INVEST DERMATOL
MENEGUZZI G, 2000, P97, SKIN GENE THERAPY
NISHIZAWA Y, 1993, V113, P493, J BIOCHEM-TOKYO
PALAZZI X, 2000, V115, P135, J INVEST DERMATOL
ROBBINS PB, 2001, V98, P5193, P NATL ACAD SCI USA
ROUSSELLE P, 1997, V138, P719, J CELL BIOL
RYAN MC, 1999, V145, P1309, J CELL BIOL
SAHLBERG C, 1998, V77, P1589, J DENT RES
SALO S, 1999, V18, P197, MATRIX BIOL
SAMBROOK J, 1989, MOL CLONING LAB MANU
SASAKI T, 2001, V314, P751, J MOL BIOL
SEITZ CS, 1999, V6, P42, GENE THER
SHAPIRO J, 1995, V36, P572, CAN VET J
SHIMIZU H, 1997, V289, P174, ARCH DERMATOL RES
SONNENBERG A, 1987, V262, P10376, J BIOL CHEM
SPIRITO F, 2001, V3, P21, J GENE MED
STAHL S, 1997, V110, P55, J CELL SCI 1
SUGIYAMA S, 1995, V228, P120, EUR J BIOCHEM
THOMPSON JD, 1994, V22, P4673, NUCLEIC ACIDS RES
UITTO J, 2001, V137, P1458, ARCH DERMATOL
VAILLY J, 1994, V219, P209, EUR J BIOCHEM
VAILLY J, 1998, V5, P1322, GENE THER
VIDAL F, 1995, V10, P229, NAT GENET
WOJCIK SM, 2001, V154, P619, J CELL BIOL .
ZENT R, 2001, V238, P289, DEV BIOL

15/9/3 (Item 1 from file: 155)
DIALOG(R)File 155:MEDLINE(R)

10188137 22218275 PMID: 12230513
Animal models for skin blistering conditions: absence of laminin 5 causes hereditary junctional mechanobullous disease in the Belgian horse.
Spirito Flavia; Charlesworth Alexandra; Linder Keith; Ortonne Jean-Paul; Baird John; Meneguzzi Guerrino
INSERM U385, Faculte de Medecine, Nice, France.
Journal of investigative dermatology (United States) Sep 2002, 119 (3) p684-91, ISSN 0022-202X Journal Code: 0426720
Document type: Journal Article

Languages: ENGLISH
Main Citation Owner: NLM
Record type: Completed
Subfile: INDEX MEDICUS

Recent achievements in the genetic correction of keratinocytes isolated from patients with junctional epidermolysis bullosa have paved the way to a gene therapy approach for the disease. Because gene therapy protocols require preclinical validation in animals, we have characterized spontaneous animal models of junctional epidermolysis bullosa. In this study we have elucidated the genetic basis of the hereditary junctional mechanobullous disease in the Belgian horse, a condition characterized by blistering of the skin and mouth epithelia, and exungulation (loss of the hoof). Immunofluorescence analysis associated the condition to the absent expression of the gamma2 chain of laminin 5 and designated Lamc2 as the candidate gene. Comparative analysis of the nucleotide sequence of the full-length gamma2 cDNA isolated by reverse transcription polymerase chain reaction amplification of total RNA purified from the epithelium of a junctional epidermolysis bullosa foal and a healthy control disclosed a homozygous basepair insertion (1368insC) in the affected animal. Mutation 1368insC results in a downstream premature termination codon and is predicted to cause absent expression of the laminin gamma2 polypeptide. Our results also show that: (i) the horse junctional epidermolysis bullosa genetically corresponds to the severe Herlitz form of junctional epidermolysis bullosa in man; (ii) the amino acid sequence and structure of the horse laminin gamma2 chain are virtually identical to the human counterpart; (iii) the moderate eruption of skin blisters in the affected animals with respect to the human Herlitz junctional epidermolysis bullosa patients correlates with the protection provided by hair. Our observations suggest that the affected foals are a convenient source of epithelial cells from tissues that cannot be obtained from human junctional epidermolysis bullosa patients, and imply that hairless strains of animals with recessive skin disorders would be the best models for in vivo gene therapy approaches to skin blistering diseases.

Tags: Animal; Human; Support, Non-U.S. Gov't
Descriptors: *Cell Adhesion Molecules--genetics--GE; *Disease Models, Animal; *Epidermolysis Bullosa, Junctional--genetics--GE; *Epidermolysis Bullosa, Junctional--physiopathology--PP; *Horses; Blister--genetics--GE; Blister--physiopathology--PP; DNA, Complementary; Epithelium--pathology --PA; Genotype; Joints--pathology--PA; Laminin--genetics--GE; Molecular Sequence Data; Pedigree; Point Mutation; Sequence Homology, Amino Acid
CAS Registry No.: 0 (Cell Adhesion Molecules); 0 (DNA, Complementary) ; 0 (Laminin); 0 (kalinin); 0 (laminin gamma 2)
Record Date Created: 20020916
Record Date Completed: 20021010

? ds

Set	Items	Description
S1	1899	"EPIDERMOLYSIS BULLOSA"
S2	48128	CYTOSINE
S3	586	1368
S4	0	?1368?
S5	700	1368?
S6	40863	LAMININ
S7	1810	S6 AND (INSERT OR MUTATION OR MUTATED OR INSERTION OR DELETION OR SUBSTITUTION)
S8	700	S3 OR S5
S9	0	S1 AND S8
S10	0	S8 AND S2
S11	37	S7 AND S1
S12	1	S11 AND CYTOSINE
S13	201	LAMC2
S14	0	S13 AND S3

S15 3 S13 AND S5
? s s1 and s2
1899 S1
48128 S2
S16 2 S1 AND S2
? type s16/full/all

16/9/1 (Item 1 from file: 5)
DIALOG(R)File 5:Biosis Previews(R)

08474655 BIOSIS NO.: 199344024655
PCR-based detection of two exonic polymorphisms in the human type VII collagen gene (COL7A1) at 3p21.1.
AUTHOR: Christiano Angela M(a); Chung-Honet Linda C; Hovnanian Alain; Uitto Jouni
AUTHOR ADDRESS: (a)Dep. Dermatol., Jefferson Med. College, Thomas Jefferson University, Philadelphia, Pa. 19107
JOURNAL: Genomics 14 (3):p827-828 1992
ISSN: 0888-7543
DOCUMENT TYPE: Article
RECORD TYPE: Citation
LANGUAGE: English
REGISTRY NUMBERS: 81295-04-7: ALUI; 73-40-5Q: GUANINE; 69257-39-2Q: GUANINE ; 73-24-5: ADENINE; 71-30-7: CYTOSINE; 60-18-4: TYROSINE
DESCRIPTORS:
MAJOR CONCEPTS: Anthropology; Biochemistry and Molecular Biophysics; Clinical Chemistry (Allied Medical Sciences); Dermatology (Human Medicine, Medical Sciences); Genetics; Pathology; Population Genetics (Population Studies)
BIOSYSTEMATIC NAMES: Hominidae--Primates, Mammalia, Vertebrata, Chordata, Animalia
ORGANISMS: Hominidae (Hominidae)
BIOSYSTEMATIC CLASSIFICATION (SUPER TAXA): animals; chordates; humans; mammals; primates; vertebrates
CHEMICALS & BIOCHEMICALS: ALUI; GUANINE; ADENINE; CYTOSINE; TYROSINE
GEOGRAPHICAL NAME: USA (North America, Nearctic region)
MISCELLANEOUS TERMS: ALLELIC FREQUENCY; ALUI POLYMORPHISM; CAUCASIAN; CO-SEGREGATION; COMPLEMENTARY DNA; CYTOSINE TO TYROSINE TRANSITION; DIAGNOSTIC METHOD; EPIDERMOLYSIS BULLOSA; FINNS; GENE MAPPING; GENE MARKER; GREEKS; GUANINE TO ADENINE TRANSITION; JAPANESE; MENDELIAN SEGREGATION; MOLECULAR DIAGNOSTICS; NOTE; POLYMERASE CHAIN REACTION; PVUII POLYMORPHISM; RESTRICTION FRAGMENT LENGTH POLYMORPHISM; SOUTHERN BLOT
CONCEPT CODES:
03508 Genetics and Cytogenetics-Human
03509 Genetics and Cytogenetics-Population Genetics (1972-)
05000 Physical Anthropology; Ethnobiology
10006 Clinical Biochemistry; General Methods and Applications
10062 Biochemical Studies-Nucleic Acids, Purines and Pyrimidines
10506 Biophysics-Molecular Properties and Macromolecules
12504 Pathology, General and Miscellaneous-Diagnostic
18506 Integumentary System-Pathology
02508 Cytology and Cytochemistry-Human
18004 Bones, Joints, Fasciae, Connective and Adipose Tissue-Physiology and Biochemistry
BIOSYSTEMATIC CODES:
86215 Hominidae

16/9/2 (Item 1 from file: 34)

DIALOG(R)File 34:SciSearch(R) Cited Ref Sci

11469061 Genuine Article#: 654YK Number of References: 22
Title: A mutation in the LAMC2 gene causes the Herlitz junctional epidermolysis bullosa (H-JEB) in two French draft horse breeds
Author(s): Milenkovic D; Chaffaux S; Taourit S; Guerin G (REPRINT)
Corporate Source: INRA,Ctr Rech Jouy, Dept Genet Anim, Lab Genet Biochim & Cytogenet,F-78352 Jouy En Josas//France/ (REPRINT); INRA,Ctr Rech Jouy, Dept Genet Anim, Lab Genet Biochim & Cytogenet,F-78352 Jouy En Josas//France/
Journal: GENETICS SELECTION EVOLUTION, 2003, V35, N2 (MAR-APR), P249-256
ISSN: 0999-193X Publication date: 20030300
Publisher: E D P SCIENCES, 7, AVE DU HOGGAR, PARC D ACTIVITES COURTABOEUF, BP 112, F-91944 LES ULIS CEDEXA, FRANCE
Language: English Document Type: ARTICLE
Geographic Location: France
Journal Subject Category: AGRICULTURE, DAIRY & ANIMAL SCIENCE; GENETICS & HEREDITY
Abstract: Epidermolysis bullosa (EB) is a heterogeneous group of inherited diseases characterised by skin blistering and fragility. In humans, one of the most severe forms of EB known as Herlitz-junctional EB (H-JEB), is caused by mutations in the laminin 5 genes. EB has been described in several species, like cattle, sheep, dogs, cats and horses where the mutation, a cytosine insertion in exon 10 of the LAMC2 gene, was very recently identified in Belgian horses as the mutation responsible for JEB. In this study, the same mutation was found to be totally associated with the JEB phenotype in two French draft horse breeds, Trait Breton and Trait Comtois. This result provides breeders a molecular test to better manage their breeding strategies by genetic counselling.
Descriptors--Author Keywords: horse ; LAMC2 ; epidermolysis bullosa ; laminin 5
Identifiers--KeyWord Plus(R): MECHANOBULLOUS DISEASE; CLASSIFICATION; DIAGNOSIS; POSITION
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DUBIELZIG RR, 1986, V23, P325, VET PATHOL
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GOUREAU JM, 1989, V62, P345, B ACAD VET FR
HOOD J, 2001, V11, P463, TRENDS CELL BIOL
JOHNSON GC, 1998, V99, P329, J COMP PATHOL
KOHN CW, 1989, V21, P297, EQUINE VET J
KORGE BP, 1996, V74, P59, J MOL MED-JMM
LYKKEANDERSEN J, 2001, V293, P1836, SCIENCE
NAGY E, 1998, V23, P198, TRENDS BIOCHEM SCI
OLIVRY T, 1999, V36, P616, VET PATHOL
PALAZZI X, 2000, V115, P135, J INVEST DERMATOL
PULKKINEN L, 1999, V18, P29, MATRIX BIOL
SPIRITO F, 2002, V3, P684, J INVEST DERMATOL
TERWILLIGER JD, 1995, V56, P777, AM J HUM GENET
? ds

Set	Items	Description
S1	1899	"EPIDERMOLYSIS BULLOSA"

S2 48128 CYTOSINE
S3 586 1368
S4 0 ?1368?
S5 700 1368?
S6 40863 LAMININ
S7 1810 S6 AND (INSERT OR MUTATION OR MUTATED OR INSERTION OR DELETION OR SUBSTITUTION)
S8 700 S3 OR S5
S9 0 S1 AND S8
S10 0 S8 AND S2
S11 37 S7 AND S1
S12 1 S11 AND CYTOSINE
S13 201 LAMC2
S14 0 S13 AND S3
S15 3 S13 AND S5
S16 2 S1 AND S2
? s (s3 or s5) and cytosine
586 S3
700 S5
48128 CYTOSINE
S17 0 (S3 OR S5) AND CYTOSINE
? s s2 and s7
48128 S2
1810 S7
S18 6 S2 AND S7
? type s18/full/all

18/9/1 (Item 1 from file: 5)
DIALOG(R)File 5:Biosis Previews(R)

09637237 BIOSIS NO.: 199598092155
A homozygous nonsense mutation in the beta-3 chain gene of laminin 5 (LAMB3) in Herlitz junctional epidermolysis bullosa.
AUTHOR: Pulkkinen Leena; Christiano Angela M; Gerecke Donald; Wagman D Wolfe; Burgeson Robert E; Pittelkow Mark R; Uitto Jouni(a)
AUTHOR ADDRESS: (a)Dep. Dermatol., Jefferson Medical College, 233 South 10th Street, Room 450, Philadelphia, PA 191**USA
JOURNAL: Genomics 24 (2):p357-360 1994
ISSN: 0888-7543
DOCUMENT TYPE: Article
RECORD TYPE: Abstract
LANGUAGE: English

ABSTRACT: Herlitz junctional epidermolysis bullosa (H-JEB) is a severe autosomal recessive disorder characterized by blister formation within the dermal-epidermal basement membrane. Based on immunofluorescence analysis recognizing laminin 5 epitopes (previously known as nicein/kalinin), the genes for this lamina lucida protein have been proposed as candidate genes in H-JEB. In this study, we examined the gene encoding the beta-3 polypeptide chain of laminin 5 (LAMB3) by Northern hybridization and RT-PCR analysis of keratinocyte mRNA from a proband in a family with H-JEB. Northern analysis revealed markedly reduced levels of the laminin beta-3 chain mRNA. Amplification of mRNA by RT-PCR, followed by direct nucleotide sequencing, revealed a homozygous C-to-T transition resulting in a premature termination codon (CGA fwdarw TGA) on both alleles. This mutation was verified at the genomic DNA level, and both parents were shown to be heterozygous carriers of the same mutation. This is the first description of a mutation in the laminin beta-3 chain gene (LAMB3) of laminin 5 in an H-JEB patient.

DESCRIPTORS:
MAJOR CONCEPTS: Biochemistry and Molecular Biophysics; Cell Biology; Dermatology (Human Medicine, Medical Sciences); Development; Genetics; Membranes (Cell Biology)
BIOSYSTEMATIC NAMES: Hominidae--Primates, Mammalia, Vertebrata, Chordata, Animalia
ORGANISMS: human (Hominidae)
BIOSYSTEMATIC CLASSIFICATION (SUPER TAXA): animals; chordates; humans; mammals; primates; vertebrates
MISCELLANEOUS TERMS: AUTOSOMAL RECESSIVE DISORDER; BASEMENT MEMBRANE; BETA-3 CHAIN; CYTOSINE-TO-THYMINE TRANSITION; KERATINOCYTE MESSENGER RNA; LAMINA LUCIDA PROTEIN
CONCEPT CODES:
02508 Cytology and Cytochemistry-Human
03508 Genetics and Cytogenetics-Human
10062 Biochemical Studies-Nucleic Acids, Purines and Pyrimidines
10064 Biochemical Studies-Proteins, Peptides and Amino Acids
10508 Biophysics-Membrane Phenomena
18506 Integumentary System-Pathology
25552 Developmental Biology-Embryology-Descriptive Teratology and Teratogenesis
BIOSYSTEMATIC CODES:
86215 Hominidae

18/9/2 (Item 1 from file: 34)
DIALOG(R)File 34:SciSearch(R) Cited Ref Sci

11469061 Genuine Article#: 654YK Number of References: 22
Title: A mutation in the LAMC2 gene causes the Herlitz junctional epidermolysis bullosa (H-JEB) in two French draft horse breeds
Author(s): Milenkovic D; Chaffaux S; Taourit S; Guerin G (REPRINT)
Corporate Source: INRA,Ctr Rech Jouy, Dept Genet Anim, Lab Genet Biochim & Cytogenet,F-78352 Jouy En Josas//France/ (REPRINT); INRA,Ctr Rech Jouy, Dept Genet Anim, Lab Genet Biochim & Cytogenet,F-78352 Jouy En Josas//France/
Journal: GENETICS SELECTION EVOLUTION, 2003, V35, N2 (MAR-APR), P249-256
ISSN: 0999-193X Publication date: 20030300
Publisher: E D P SCIENCES, 7, AVE DU HOGGAR, PARC D ACTIVITES COURTABOEUF, BP 112, F-91944 LES ULIS CEDEXA, FRANCE
Language: English Document Type: ARTICLE
Geographic Location: France
Journal Subject Category: AGRICULTURE, DAIRY & ANIMAL SCIENCE; GENETICS & HEREDITY
Abstract: Epidermolysis bullosa (EB) is a heterogeneous group of inherited diseases characterised by skin blistering and fragility. In humans, one of the most severe forms of EB known as Herlitz-junctional EB (H-JEB), is caused by mutations in the laminin 5 genes. EB has been described in several species, like cattle, sheep, dogs, cats and horses where the mutation, a cytosine insertion in exon 10 of the LAMC2 gene, was very recently identified in Belgian horses as the mutation responsible for JEB. In this study, the same mutation was found to be totally associated with the JEB phenotype in two French draft horse breeds, Trait Breton and Trait Comtois. This result provides breeders a molecular test to better manage their breeding strategies by genetic counselling.
Descriptors--Author Keywords: horse ; LAMC2 ; epidermolysis bullosa ; laminin 5
Identifiers--KeyWord Plus(R): MECHANOBULLOUS DISEASE; CLASSIFICATION; DIAGNOSIS; POSITION
Cited References:

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GOUREAU JM, 1989, V62, P345, B ACAD VET FR
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OLIVRY T, 1999, V36, P616, VET PATHOL
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TERWILLIGER JD, 1995, V56, P777, AM J HUM GENET

18/9/3 (Item 2 from file: 34)
DIALOG(R)File 34:SciSearch(R) Cited Ref Sci

05982782 Genuine Article#: XM195 Number of References: 34
Title: Predominance of the recurrent mutation R635X in the LAMB3 gene in European patients with Herlitz junctional epidermolysis bullosa has implications for mutation detection strategy
Author(s): Pulkkinen L; Meneguzzi G; McGrath JA; Xu Y; BlanchetBardon C; Ortonne JP; Christiano AM; Uitto J (REPRINT)
Corporate Source: THOMAS JEFFERSON UNIV,JEFFERSON MED COLL, DEPT DERMATOL & CUTANEOUS BIOL, 233 S 10TH ST/PHILADELPHIA//PA/19107 (REPRINT); THOMAS JEFFERSON UNIV,JEFFERSON MED COLL, DEPT DERMATOL & CUTANEOUS BIOL/PHILADELPHIA//PA/19107; KUOPIO UNIV HOSP,DIV DIAGNOST SERV, CHROMOSOME & DNA LAB/SF-70210 KUOPIO//FINLAND/; THOMAS JEFFERSON UNIV,JEFFERSON MED COLL, DEPT BIOCHEM & MOL PHARMACOL/PHILADELPHIA//PA/19107; THOMAS JEFFERSON UNIV,JEFFERSON INST MOL MED, MOL DERMATOL SECT/PHILADELPHIA//PA/19107; UNIV NICE,FAC MED, INSERM, U385/NICE//FRANCE/; HOP ST LOUIS,CLIN MALAD CUTANEES/PARIS//FRANCE/; HOP ST LOUIS,UNITE RECH DIAGNOST ANTENATAL DERMATOL/PARIS//FRANCE/
Journal: JOURNAL OF INVESTIGATIVE DERMATOLOGY, 1997, V109, N2 (AUG), P 232-237
ISSN: 0022-202X Publication date: 19970800
Publisher: BLACKWELL SCIENCE INC, 350 MAIN ST, MALDEN, MA 02148
Language: English Document Type: ARTICLE
Geographic Location: USA; FINLAND; FRANCE
Subfile: CC LIFE--Current Contents, Life Sciences; CC CLIN--Current Contents, Clinical Medicine
Journal Subject Category: DERMATOLOGY & VENEREAL DISEASES
Abstract: Junctional forms of epidermolysis bullosa (JEB) are characterized by tissue separation at the level of the lamina lucida, We have recently disclosed specific mutations in the LAMA3, LAMB3, and LAMC2 genes encoding the subunit polypeptides of the anchoring filament protein laminin 5 in 66 families with different variants of JEB, Examination of the JEB mutation database revealed recurrence of a particular C-->T substitution at nucleotide position 1903 (exon 14) of LAMB3, resulting in the mutation R635X. The inheritance of

this nonsense mutation was noted on different genetic backgrounds, suggesting that R635X is a hotspot mutation, In this study, we have performed mutation evaluation in a European cohort of 14 families with the lethal, Herlitz type of JEB (H-JEB), The families were first screened for the presence of the R635X mutation by restriction enzyme digestion of the PCR product corresponding to exon 14. Four of the probands were found to be homozygous and six were heterozygous for R635X. The remaining alleles were subjected to mutation screening by PCR amplification of individual exons of LAMB3 and LAMC2, followed by heteroduplex analysis and nucleotide sequencing, In three families (six alleles), mutations in LAMC2 were disclosed, In the remaining eight alleles, additional pathogenetic LAMB3 mutations were found, None of the patients had LAMA3 mutation, Thus, LAMB3 mutations accounted for 22 of 28 JEB alleles (79%), and a total of 14 of 22 LAMB3 alleles (64%) harbored the R635X mutation, signifying its prevalence as a predominant genetic lesion underlying H-JEB in this European cohort of patients, This recurrent mutation will facilitate screening of additional JEB patients for the purpose of prenatal testing of fetuses at risk for recurrence.

Descriptors--Author Keywords: basement membrane zone ; laminin 5 mutations
Identifiers--KeyWord Plus(R): HOMOZYGOUS NONSENSE MUTATION; BETA-3 CHAIN GENE; LAMININ-5 LAMB3; VII COLLAGEN
Research Fronts: 95-0068 002 (DYSTROPHIN GENE; SARCOGLYCAN COMPLEX; MDX MUSCLE)
95-0857 001 (AORTIC DISSECTION; TRANSESOPHAGEAL ECHOCARDIOGRAPHY; NEONATAL MARFAN-SYNDROME)
95-4533 001 (P53 GENE; SPONTANEOUS HPRT MUTATIONS; CPG CYTOSINE METHYLATION; HOMOZYGOUS FAMILIAL HYPERCHOLESTEROLEMIA; BASE EXCISION-REPAIR; HPAII METHYLTRANSFERASE)
95-5061 001 (STRUCTURAL GENE; GLTC-DEPENDENT REGULATION OF BACILLUS-SUBTILIS GLUTAMATE SYNTHASE EXPRESSION; ARABIDOPSIS TYPE-1 PROTEIN PHOSPHATASE)

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18/9/4 (Item 3 from file: 34)
DIALOG(R)File 34:SciSearch(R) Cited Ref Sci

05827852 Genuine Article#: XA243 Number of References: 12
Title: A recurrent laminin 5 mutation in British patients with lethal (Herlitz) junctional epidermolysis bullosa: Evidence for a mutational hotspot rather than propagation of an ancestral allele
Author(s): Ashton GHS; Mellerio JE; Dunnill MGS; Pulkkinen L; Christiano AM ; Uitto J; Eady RAJ; McGrath JA (REPRINT)
Corporate Source: UNITED MED & DENT SCH GUYS & ST THOMAS HOSP,ST THOMAS HOSP, LAMBETH PALACE RD/LONDON SE1 7EH//ENGLAND/ (REPRINT); UNITED MED & DENT SCH,ST THOMAS HOSP, ST JOHNS INST DERMATOL/LONDON SE1 7EH//ENGLAND/; THOMAS JEFFERSON UNIV,JEFFERSON MED COLL, DEPT DERMATOL & CUTANEOUS BIOL/PHILADELPHIA//PA/19107; COLUMBIA UNIV,DEPT DERMATOL/NEW YORK//NY/
Journal: BRITISH JOURNAL OF DERMATOLOGY, 1997, V136, N5 (MAY), P674-677
ISSN: 0007-0963 Publication date: 19970500
Publisher: BLACKWELL SCIENCE LTD, OSNEY MEAD, OXFORD, OXON, ENGLAND OX2 0EL
Language: English Document Type: ARTICLE
Geographic Location: ENGLAND; USA
Subfile: CC LIFE--Current Contents, Life Sciences; CC CLIN--Current Contents, Clinical Medicine;
Journal Subject Category: DERMATOLOGY & VENEREAL DISEASES
Abstract: The three genes (LAMA3, LAMB3 and LAMC2) that encode the anchoring filament protein, laminin 5, may all harbour pathogenetic mutations in the autosomal recessive blistering skin disorder, junctional epidermolysis bullosa (JEB). Recently, one particular mutation, R635X in the LAMB3 gene, has been found to account for approximately 40% of all JEB laminin 5 mutations (Kivirikko et al., Hum Mol Genet 1996; 5: 231-7). In this study, we assessed the frequency of this mutation in 12 British patients with lethal (Herlitz) JEB using PCR amplification of genomic DNA and restriction endonuclease digestion. The mutation R635X was found in seven of 24 (29%) mutant alleles, confirming its relative frequency within the British gene pool. In addition, haplotype analysis using intragenic polymorphisms showed that the mutation arose on at least four different haplotype backgrounds, suggesting it represents a mutational hotspot rather than propagation of a common British ancestral allele. These findings support the hypermutable nature of this CpG dinucleotide and have implications in screening for laminin 5 gene mutations in British and other patients with JEB.
Identifiers--KeyWord Plus(R): DIAGNOSIS; GENE
Research Fronts: 95-0068 001 (DYSTROPHIN GENE; SARCOGLYCAN COMPLEX; MDX MUSCLE)
95-4533 001 (P53 GENE; SPONTANEOUS HPRT MUTATIONS; CPG CYTOSINE METHYLATION; HOMOZYGOUS FAMILIAL HYPERCHOLESTEROLEMIA; BASE EXCISION-REPAIR; HPAII METHYLTRANSFERASE)
95-5061 001 (STRUCTURAL GENE; GLTC-DEPENDENT REGULATION OF BACILLUS-SUBTILIS GLUTAMATE SYNTHASE EXPRESSION; ARABIDOPSIS TYPE-1 PROTEIN PHOSPHATASE)
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18/9/5 (Item 4 from file: 34)

05702502 Genuine Article#: WR230 Number of References: 28
Title: Fabry disease: Thirty-five mutations in the alpha-galactosidase A gene in patients with classic and variant phenotypes
Author(s): Eng CM (REPRINT) ; Ashley GA; Burgert TS; Enriquez AL; DSouza M; Desnick RJ
Corporate Source: CUNY MT SINAI SCH MED,DEPT HUMAN GENET, BOX 1498, 1 GUSTAVE LEVY PL/NEW YORK//NY/10029 (REPRINT); CUNY MT SINAI SCH MED,DEPT PEDIAT/NEW YORK//NY/10029
Journal: MOLECULAR MEDICINE, 1997, V3, N3 (MAR), P174-182
ISSN: 1076-1551 Publication date: 19970300
Publisher: SPRINGER VERLAG, 175 FIFTH AVE, NEW YORK, NY 10010
Language: English Document Type: ARTICLE
Geographic Location: USA
Subfile: CC LIFE--Current Contents, Life Sciences; CC CLIN--Current Contents, Clinical Medicine
Journal Subject Category: BIOCHEMISTRY & MOLECULAR BIOLOGY; MEDICINE, RESEARCH & EXPERIMENTAL; CELL BIOLOGY
Abstract: Background: Fabry disease, an X-linked inborn error of glycosphingolipid catabolism, results from mutations in the alpha-galactosidase A (alpha-Gal A) gene located at Xq22.1. To determine the nature and frequency of the molecular lesions causing the classical and milder variant Fabry phenotypes and for precise carrier detection, the alpha-Gal A lesions in 42 unrelated Fabry hemizygotes were determined. Materials and

Methods: Genomic DNA was isolated from affected probands and their family members. The seven alpha-galactosidase A exons and flanking intronic sequences were PCR amplified and the nucleotide sequence was determined by solid-phase direct sequencing.

Results: Two patients with the mild cardiac phenotype had missense mutations, I91T and F113L, respectively. In 38 classically affected patients, 33 new mutations were identified including 20 missense (M1T A31V, H46R, Y86C, L89P, D92Y, C94Y, A97V, R100T, Y134S, G138R, A143T, S148R, G163V, D170V, C202Y, Y216D, N263S, W287C, and N298S), two nonsense (Q386X, W399X), one splice site mutation (IVS4 + 2T --> C), and eight small exonic insertions or deletions (304del1, 613del9, 777del1, 1057del2, 1074del2, 1077del1, 1212del3, and 1094ins1), which identified exon 7 as a region prone to gene rearrangements. In addition, two unique complex rearrangements consisting of contiguous small insertions and deletions were found in exons 1 and 2 causing L45R/H46S and L120X, respectively.

Conclusions: These studies further define the heterogeneity of mutations causing Fabry disease, permit precise carrier identification and prenatal diagnosis in these families, and facilitate the

identification of candidates for enzyme replacement therapy.
Identifiers--KeyWord Plus(R): A-GENE; NUCLEOTIDE-SEQUENCE; ATYPICAL VARIANT; ALPORT SYNDROME; IDENTIFICATION; CDNA; REARRANGEMENTS; HEMIZYGOTES; MUTAGENESIS; EXPRESSION
Research Fronts: 95-1418 002 (TYPE-IV COLLAGEN ALPHA-5 CHAIN GENE (COL4A5); AUTOSOMAL RECESSIVE ALPORT SYNDROME; RENAL GLOMERULUS OF MICE LACKING S-LAMININ LAMININ BETA-2)
95-0369 001 (PLECKSTRIN HOMOLOGY DOMAINS; HETEROTRIMERIC G-PROTEINS; CLONED PLANT K+ CHANNEL IN XENOPUS OOCYTES; X-LINKED AGAMMAGLOBULINEMIA; CELLULAR EXPRESSION)
95-4533 001 (P53 GENE; SPONTANEOUS HPRT MUTATIONS; CPG CYTOSINE METHYLATION; HOMOZYGOUS FAMILIAL HYPERCHOLESTEROLEMIA; BASE EXCISION-REPAIR; HPAII METHYLTRANSFERASE)
95-5061 001 (STRUCTURAL GENE; GLTC-DEPENDENT REGULATION OF BACILLUS-SUBTILIS GLUTAMATE SYNTHASE EXPRESSION; ARABIDOPSIS TYPE-1 PROTEIN PHOSPHATASE)
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18/9/6 (Item 1 from file: 155)
DIALOG(R)File 155:MEDLINE(R)

14561668 22520922 PMID: 12633536
A mutation in the LAMC2 gene causes the Herlitz junctional epidermolysis bullosa (H-JEB) in two French draft horse breeds.
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Genetics, selection, evolution. - GSE (France) Mar-Apr 2003, 35 (2) p249-56, ISSN 0999-193X Journal Code: 9114088
Document type: Journal Article
Languages: ENGLISH

Main Citation Owner: NLM
Record type: In Process
Subfile: INDEX MEDICUS

Epidermolysis bullosa (EB) is a heterogeneous group of inherited diseases characterised by skin blistering and fragility. In humans, one of the most severe forms of EB known as Herlitz-junctional EB (H-JEB), is caused by mutations in the laminin 5 genes. EB has been described in several species, like cattle, sheep, dogs, cats and horses where the mutation , a cytosine insertion in exon 10 of the LAMC2 gene, was very recently identified in Belgian horses as the mutation responsible for JEB. In this study, the same mutation was found to be totally associated with the JEB phenotype in two French draft horse breeds, Trait Breton and Trait Comtois. This result provides breeders a molecular test to better manage their breeding strategies by genetic counselling.

Record Date Created: 20030313

? type s18/full/allds
>>>'ALLDS' not recognized as item list
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Set	Items	Description
S1	1899	"EPIDERMOLYSIS BULLOSA"
S2	48128	CYTOSINE
S3	586	1368
S4	0	?1368?
S5	700	1368?
S6	40863	LAMININ
S7	1810	S6 AND (INSERT OR MUTATION OR MUTATED OR INSERTION OR DELETION OR SUBSTITUTION)
S8	700	S3 OR S5
S9	0	S1 AND S8
S10	0	S8 AND S2
S11	37	S7 AND S1
S12	1	S11 AND CYTOSINE
S13	201	LAMC2
S14	0	S13 AND S3
S15	3	S13 AND S5
S16	2	S1 AND S2
S17	0	(S3 OR S5) AND CYTOSINE
S18	6	S2 AND S7